MORE THAN YOU CAN IMAGINE

an anthology of
rare experiences

GENETIC ALLIANCE UK

ANTHOLOGY CREATORS

Members of the genetic, rare and undiagnosed conditions community and the Rare Disease Day Team, Genetic Alliance UK.

DISCLAIMER

Any views or opinions expressed in this anthology are solely those of the individual contributors from the rare conditions community and do not necessarily represent the views or opinions of Genetic Alliance UK. Additionally, inclusion of content in this anthology is not an endorsement by Genetic Alliance UK.

FUNDING STATEMENT

This anthology was funded by our Rare Disease Day 2025 project and delivered by the Genetic Alliance UK team.

The project was funded by contributions from:

Alexion Pharmaceuticals
Amicus Therapeutics
BioCryst
Biogen
Chiesi
Jazz Pharmaceuticals
Johnson & Johnson
Kyowa Kirin
Novartis Pharmaceuticals
Pfizer
PTC Therapeutics
Sanofi
Takeda
Vertex Pharmaceuticals

All funding received is in accordance with Genetic Alliance UK's working with the life sciences industry policy.

The project is funded in line with Genetic Alliance UK's working with the **life sciences industry policy: bit.ly/42w0XXV**. Funders have had no involvement in the development of the anthology or the wider Rare Disease Day 2025 project.

Front cover illustration and back cover poem provided by Matilda Tumim - instagram.com/tumimandprendergast

About Genetic Alliance UK

Genetic Alliance UK is an alliance of over 200 charities and support groups working together to improve the lives of people in the UK with lifelong and complex genetic, rare and undiagnosed conditions.

We advocate for fast and accurate diagnosis, good quality care and access to the best treatments. We actively support progress in research and engage with decision makers and the public about the challenges faced by our community.

We run two long standing projects:

RARE DISEASE UK

A campaign focused on making sure the UK Rare Diseases Framework is as successful as possible, and to ensure that people and families living with rare conditions have access to a final diagnosis, coordinated care and specialist care and treatment.

SWAN UK

The only dedicated support network in the UK for families affected by a syndrome without a name – a genetic condition so rare it often remains undiagnosed.

'More than you can imagine: an anthology of rare experiences' is a compilation of individual creative works submitted by members of the genetic, rare and undiagnosed communities.

All contributors submitted their works on a voluntary basis. We are very grateful to everyone who has contributed to this anthology.

Please reference this publication as More than you can imagine: an anthology of rare experiences, Genetic Alliance UK (2025)

Published: 27 February 2025

Introduction

This introduction is deliberately short because we want to make sure that you can get to the good stuff, our brilliant anthology submissions, without delay.

Our aim in creating this anthology is to amplify the voices of the genetic, rare and undiagnosed communities to improve understanding of the impact of living with rare conditions. We also want these collected creative expressions to inform and influence the public, healthcare professionals, policy makers, parliamentarians and the wider community.

Genetic Alliance UK is uniquely positioned to extend the reach and impact of these experiences and we hope that you are as profoundly affected by the Anthology's content as we have been.

We can't overstate how honoured, pleased and proud we are to share this collection of creative works with you for Rare Disease Day 2025. We were blown away by both the quality and quantity of submissions and we are more grateful than you can imagine to our rare community for their generosity and for trusting us with their experiences.

The Genetic Alliance UK Team

Support

If you've been affected by anything in this anthology then support is available.

Genetic Alliance UK's website lists crisis support providers:

geneticalliance.org.uk/support-and-information/crisis-support

Rareminds Wellbeing Hub has lots of resources and information for people affected by genetic, rare and undiagnosed conditions:

rareminds.org/wellbeing-hub

More than you can imagine

Anthony Heard, Reading
Immune thrombocytopenia and fibromyalgia

'This is about living with ITP for nearly 19 years now. The uncertainty of it, the shock of it and the stress of never ending tests, appointments. It also highlights the importance of having the support of a strong rare disease community.'

More than you can imagine
Living a life of rare
Being diagnosed in 2006
Almost more than I could bear

More than I could imagine
The appointments, tests and scans
Almost a total wipeout
Of our carefully constructed plans

More than you can imagine
The shock of ITP
So many times I've asked myself
How did it happen to me ?

More than I could imagine
The uncertainty and the strain
Not just the fatigue or bruising
The physical and mental pain

More than you can imagine
It's far from the best fun I've had
But when compared to others
I'm grateful things aren't so bad

More than I could imagine
300 million of us Worldwide
It's just so wonderful to know
There are so many on my side

More than you can imagine
A day set aside for rare
It's incumbent on every one of us
To make other people aware

More than I can imagine
Not at all, it's a hurdle on the way
We all have challenges & crosses to bear
So finally all I would say

is –

NONE OF US CAN DO EVERYTHING BUT WE CAN ALL DO SOMETHING.

WE HAVE TO BE THE CHANGE WE SEEK.

PLEASE JOIN IN WITH RARE DISEASE DAY FEB 28TH '25.

More plastic than you could ever imagine

Joanne Williams, Birmingham
Undiagnosed life-limiting neurogenetic condition

‘Being a parent to a medically complex child thrusts you into a world you had never imagined. Overnight your role changes from parent to nurse/physio/dietician/advocate and first responder. The enormity of responsibility is overwhelming. In creating this necklace I wanted to present a sample of the physical volume of medical paraphernalia and illustrate the unseen emotional weight of worry and grief involved in managing my son’s condition on a daily basis. I have a love/hate relationship with these items because of what they represent in our life but also wanted to honour them in a weird way.

My son has an as yet undiagnosed life-limiting neurogenetic condition (a link to a new gene is currently being explored). He has significant malformations of the corpus callosum with associated conditions such as epilepsy, dystonia, dyskinesia and cerebral irritability. His needs are profound and complex. He is non verbal and it is unclear whether he has any visual function. He is PEG fed and requires a wheelchair to move around.

Reuben is our youngest child and one of three children. It is important to know that our journey into the world of medical parenting began in 2018 when our eldest son Ben was diagnosed with an aggressive brain tumour. He sadly passed away in May 2019, 11 months before Reuben was born. Ben was six years old and had previously been healthy. The two conditions are unrelated.’

Will I survive?

David Wilson, Stokesley
Ehlers Danlos syndrome

‘This poem was written in hospital just a few months ago as I was being treated for a rare life-threatening condition resulting from my Ehlers Danlos syndrome.’

Will I survive
This thing inside of me
This ticking bomb
This fault of tangled genes?

Can I hope in surgeons’ skill
Or sheer good luck
To turn the odds
Defy the fatal flaw once more?

For now I walk
With painful gait
Bask in the sun and feel
The gentle breeze caress my skin

Will I survive
With fierce hope
Buoyed by love and care
And joy and prayer

More noises than you can imagine

Audrey Harris, England
Marfan syndrome

‘My daughter was in intensive care prior to her diagnosis with Marfan syndrome. She was 11 at the time and I wrote this poem when I was sat by her bed.’

Bleep for the water
Bleep for the air
Bing for medication
Keeping her here

Buzz for the nurse
In silence alone
Crash it’s the other kids
The ones going home

Knock it’s the doctors
Tap for teacher
In comes the physio
Go away Mr preacher

Trill it’s the ventilator
Creak it’s the bed
Bong go the SATS
All the sounds in my head

It’s the song of the hospital
I will forever hear
The noises of healing
The noises of fear

Pills, pills and more pills!

Louise, Edinburgh
Fanconi anaemia
Submitted by Robert Dalgleish

‘Louise created an image of a patient lying on a hospital bed made out of her pill boxes. Created shortly after her bone marrow transplant, aged 12, when her daily routine was just “pills, pills, and pills”.’

The call to be brave

Becky Bell, Stone
Microphthalmia

'This piece of work is dedicated to the individuals who instilled in me the courage to confront life's challenges and taught me how to live with a disability – my parents.'

More life threatening than you can imagine

Natalie Robertson, Eastleigh
Thrombotic thrombocytopenic purpura (TTP)

This is life threatening,
'Treatment within 4 hours' they say,
'We don't know if you will make it,
But we want you to stay',

This is life threatening,
I'm too young to die,
What do I say to my mum?
How do I say goodbye?

This is life threatening,
And extremely rare,
Why is this happening to me?
How is this fair?

This is life threatening,
With family by my side,
I'm so weak and tired,
Can I go outside?

This is life threatening,
I can't even wash my hair,
Where has all my energy gone,
Please, send a prayer

This is life threatening,
There is no cure,
This my life now,
I'm so insecure

This is life threatening,
Am I going to relapse,
Am I going to go through
this again?
I think I might collapse

More than you can imagine

Gillian Rich, Nottingham
Primary biliary cholangitis

'I was diagnosed with PBC twenty years ago and this submission takes one through my journey from isolation to love, joy and support. I have been a member of the PBC Foundation, working with Wendy to raise awareness of the condition for the last ten years.'

Let me take you on a journey of two very different halves and how that journey has affected me - more than you can imagine.

2004 - hit with a diagnosis of Primary Biliary Cholangitis [PBC].
It is rare. It is chronic. It is an autoimmune condition. It is invisible. It is the liver.

Let your imagination now take you through winds and rains
As I feel my aches and pains
Then come with me travelling north to south
And feel the dryness of my mouth
Imagine winter chills make me quiver
That's me living with my poorly liver
Then night times dreams make me twitchy
Followed by continually being itchy
Imagine your mouth and lips feeling on fire
I am struggling to eat with no saliva
Then fatigue hits halfway through my dialogue
As time stands still ----- brain fog
This is probably more than you can imagine
To live with my ten years of isolation
Ten years of telling no one
[well, it is a liver complaint]
Ten years of "why me?"
Ten years of looking good [really]
So, ten years of knowing no one will understand
Not even me.

Hand on heart, it is more than you can imagine.
That first hug; the magic; the love; the laughter. No more isolation. Someone knows how I am really feeling. Words did not have to be spoken. We understand each other immediately. Those hugs over these ten years have remained so special.
So, trust me when I say, the feeling is so much more than you can imagine. Take my word for it. The power of the hug, of a smile, of a touch, is even more than you can imagine.
So, it must be hard for you to imagine that I am glad that I have got PBC. In the last ten years, I must have hugged almost 50 of the 20,000 PBC folk in the world. IMAGINE THAT. IMAGINE THE HUGS.

Life with Williams syndrome is more than you can imagine...

Liz Martin, Burton-On-Trent
Williams syndrome

I was so excited to have a baby, with a heart so full of dreams
Not knowing that they would soon be ripped apart at the seams…
I was broken as each night my baby cried and cried,
She would not lay down and sleep, no matter what I tried.
I was told that I was neurotic, it was all in my head,
While sleep deprivation left me frayed, hanging by a thread.
I needed answers - to know why, and what, was happening -
The dismissals, more frustrating than you can imagine…

When my little girl was almost three; answers were posted through my door
Williams Syndrome was the reason - I hadn't heard of it before.
With weeks until an appointment, I turned to Google to read
About the things she would miss out on, and the lifelong care she'd need.
She wouldn't have friends, or a family of her own,
She wouldn't have a job or manage her own home.
I was lost and alone, I didn't know where to begin
Being rare, is more isolating that you can imagine…

The appointments poured in, and a common theme was set -
She was the first with Williams most professionals had met.
At her heart scan, she wriggled - she was surprisingly strong!
The cardiologist tutted at me - what had I done wrong..?
Next, I needed to learn how to advocate and fight
For the educational support that was her legal right.
There were so many meetings, and forms to fill in,
The responsibility was heavier than you can imagine…

We needed to find others sharing our situation
So, we signed up and joined the Williams Syndrome Foundation.
We went to our first meet up, and have never looked back…
We soon became a family, it was as simple as that.
We have supported each other over life's many hurdles
And reaped the benefits of Williams with the abundance of cuddles!
Despite all the challenges, we consider Williams as a blessing,
Having Evie has made our lives richer than you can imagine…

More pain than you could imagine

Lauren Jennings, South Benfleet
Sacrococcygeal teratoma

'Sacrococcygeal teratoma – removed in 2019 and another removed in 2020. Bowel repair needed and 3 major surgeries. I now suffer with chronic pain, chronic fatigue and need a walking stick to get around.'

More uncertain than you can imagine

Hayley Harrison, Derbyshire
Undiagnosed condition

I watched you, noticed things and
began to raise concern.
"Don't compare your baby to
others" is something I really tried
to learn.

But what about those
unreachable milestones, the ones
you didn't meet?
The way you couldn't stand or
balance upon your tiny feet.

Flagging up these worries, being
told it's 'just what babies do'.
But knowing in my heart there
was something more behind
it too.

Referrals began, a process started.
At last we were being heard.
But as a result of these referrals
came letters and discussions,
I can remember every word…

Reading and talking about some
unfathomable outcomes,
Tests for atrophy and distrophy.
It's in black and white in those
letters and still now it causes such
trauma for me.

Blood samples taken, screams
and tears were shed from not just
you but me.
As your mummy watching you go
through these things fed fierce
health anxiety.

How can my world, my pride
and joy, suddenly feel so
misunderstood now?
Being passed from consultant to
consultant for answers hoping
each will tell us now.

I'll never forget the moments in
appointments, unable to calm my
racing heart.
Holding back tears and taking
a few deep breaths when asked
each time "talk us through from
the start."

Physiotherapy to orthopaedics,
paediatrics to neurology
Taking you along each time, not
understanding what's happening,
just didn't seem fair to me.

"He's a mystery to me" were the
words the neurologist spoke
when discussing what he knew.
"We'll do a brain and spine MRI,
more blood tests and a nerve
conduction test too."

I'll never forget the feeling of
watching you writhing in
utter fear,
As we restrained you to put
you to sleep with the mask
Repeating "I love you"
in your ear.

You woke confused, upset
beyond words,
A panic attack in full grasp.
At least we might have some
answers now,
How naive to think it would be
done at last.

Another appointment, this is it
I thought, we're finally going
to know.
Why you need a walking frame,
splints and help to move, just to
get up and go.

The results were inconclusive but
a Syrinx was found in
your spine.
Another spinal x-ray done to
check if all is fine.

Wheelchair services then involved,
with waiting lists galore.
A wheelchair finally allocated
To help your tired legs explore.

Helping my son meeting physical
targets, supporting him to shine.
The plan is to watch, wait and
repeat the tests,
Being told we need to give it
more time.

Time…

Time to wait.
Time to wonder.
Time to worry.
Time to question.

Time is lonely.
Time is confusing.
Time is isolating.
Time stretches out the
milestone gaps even more
between you and your peers.
Time brings new
accomplishments but greater
challenges and fears.

It will take time.

Time could be years.
There may never be an answer
after all of this time.
A Syndrome Without a Name
is what it might remain after
all of this time.

My son
My swan.

There is one thing of certainty
that I do know through all of this
process, through all of this time.

We will face this uncertain time
together and I'm so thankful that
you are mine.

More paperwork than you can imagine

Natalie Parr, Coventry,
Guillain-Barré syndrome

'I have 24 hour carers and find forms so difficult it's not me who does them on my own, someone does it. But I never fit in any boxes, I am always having to add extra pages and we always seem to have so many forms. Sometimes it's the paperwork that comes with the medical conditions that is so so draining! The CHC forms, PIP forms and everything and there is so much paperwork. But there's never enough space to write the list of meds or anything. Explaining can be draining!!'

"A little form of boxes to tick for us please"
at a glance I can see, this will be a squeeze!
And try as I might mine just won't fit in...
Let me explain... oh where to begin

On paper the tick list asks what is wrong
The tick box is tiny, the diagnosis name is long
"Please write your medicines", there's one box to fill
More medicines than you imagine not just one trusty pill!!

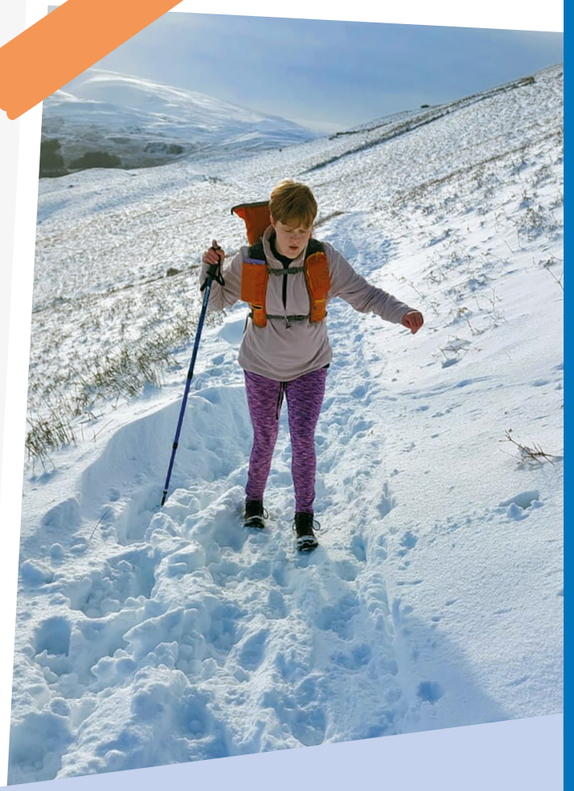
"Please write your Doctor and treatment below"
Five hospitals, many doctors... the words overflow
"Please write down important things of note"
A list that is huge won't fit in one quote!

"Do you suffer from any of these things named here"
No space to expand or explain or be clear
I'm rare and I'm "complex" but living life in full glory
But I don't fit a tick box... a box can't fit my whole story!

Please see attached sheet, my 52 meds are noted
More paper than you imagine... your form now so bloated!
Please see attached sheet for the summary of me...
Explaining explaining is so draining you see!

I don't fit your tick box, but I still love how I'm living
I'm human and caring, I'm kind and I'm giving
But I'm judged as so complex, disabled and ill
I'm costly and draining to the NHS bill

But I still want to live in a community close knit
Not rammed in a tick box I simply don't fit
Where society judges me for being so rare
I just want a chance to be treated as fair



I'm more adventurous than you might imagine

Clare Millington, Carnforth
DDX3X syndrome

'I am mother to identical twins with DDX3X syndrome. It is de novo – I do not have it myself. Although DDX3X syndrome is characterised by developmental delays, learning disability and autism, I still want to push the boundaries of what is possible. I need physical challenges to calm my over-active sensory needs.'

More questions than you can ever imagine

Julie Clayton, Stafford
Pseudo xanthoma elasticum

My life changed
When we were married.
Not for the reasons you think!
I was diagnosed with PXE.

What on earth is it?
I did not know, I did not care.
I was somewhere beautiful,
With my new husband.

But then it changed,
I had decisions to make.
Nobody had knowledge
Of the disease I lived with.

They test me repeatedly,
Invite trainees to watch.
Nobody gives me answers or understands
The disease affecting my body.

I've been told I'll die early.
I've been told anything might happen,
I've been told it could be your PXE,
Every time I have a problem.

I'm tired and drained,
I have nobody to help me.
Nobody understands,
not professionals or family.

I'm a human guinea pig,
Tested all of the time,
But this is for their sake,
Never for mine!

I am the guinea pig
Everyone mentions
No solution, no guidance.
Just watch and see!

I don't want to do this!
I don't want to be prodded!
I don't want to be tested!
I just want some care!

More than you can ever imagine.

Surrender

Robyn Gunner, Swaffham
Muscular dystrophy

'I was battling with depression, after losing care from social services due to cutbacks and having been verbally abused by a bus driver, due to me needing the ramp to access the bus! Hospital visits were always just so difficult with lack of education on Muscular dystrophy and lack of wanting to care from staff. The person in the wheelchair is waving a white flag to surrender, depicting I couldn't take anymore. Above is a bomb with UK flag, as I feel government and council do not care about the disabled. Below is fire, society's treatment towards disabilities.'

More ironic than you can imagine

Mhairi Hastie, Longniddry
Kallmann's syndrome, part hypogonadotropic hypogonadism

Born without a sense of smell is one symptom of the rare and complicated condition of Kallmann's Syndrome but the one that causes the most intrigue amongst non-sufferers. KS is part of Hypogonadotropic Hypogonadism. It affects hormonal growth and bone density which means those who manage KS /CHH have to navigate adolescence and beyond, without going through puberty. It's not life threatening but hugely life affecting and the mental impact enormous. There are physical defects often not immediately visible, and its rarity means that no one has ever heard of it. It is not something I readily tell folk about, through fear and guilt, but my older self realises the way forward is to raise awareness.

After the initial reaction of disbelief, it is beyond comprehensible by some how anyone can cope and survive in the ever-changing sensory world without this crucial sense. And indeed, it has caused me a few scary moments, but a fair few amusing moments too. But I have four other senses which I can use and as I have no experience of life with a sense of smell, it is totally normal. To me the mental and physical horrors endured not going through puberty, the effects of non-hormonal development and my lack of physical development far outweighs any concern I have over my anosmia. It is just one aspect, and KS seems to present itself differently in each individual.

In raising awareness, I have discovered understanding of the real-life affecting experiences of living with a rare condition, is based on what is relatable. But it is raising awareness.

Smell seems to be relatable and a life without it, unimaginable, yet it is well beyond my imagination. More ironic than people can imagine!

More pronunciations than you can imagine

*Reynard the Fox versus Raynaud the Physician**

Viv Sayer, Carmarthen
Antisynthetase syndrome

'As someone with a background in English Lit, teaching and editing, l am amused/infuriated when even my clinician mispronounces Raynaud's! Hence the Villanelle which contrasts the original French with the most common mispronunciation, which happens to be the name of a fox who appears in medieval fables!'

Three cheers for the Frenchman called Maurice Raynaud,
Who discovered a blood-flow condition,
But Reynard's the fox who can still steal the show!

If your fingers turn white when the temperature's low
You will need to consult a physician:
Three cheers for the Frenchman called Maurice Raynaud.

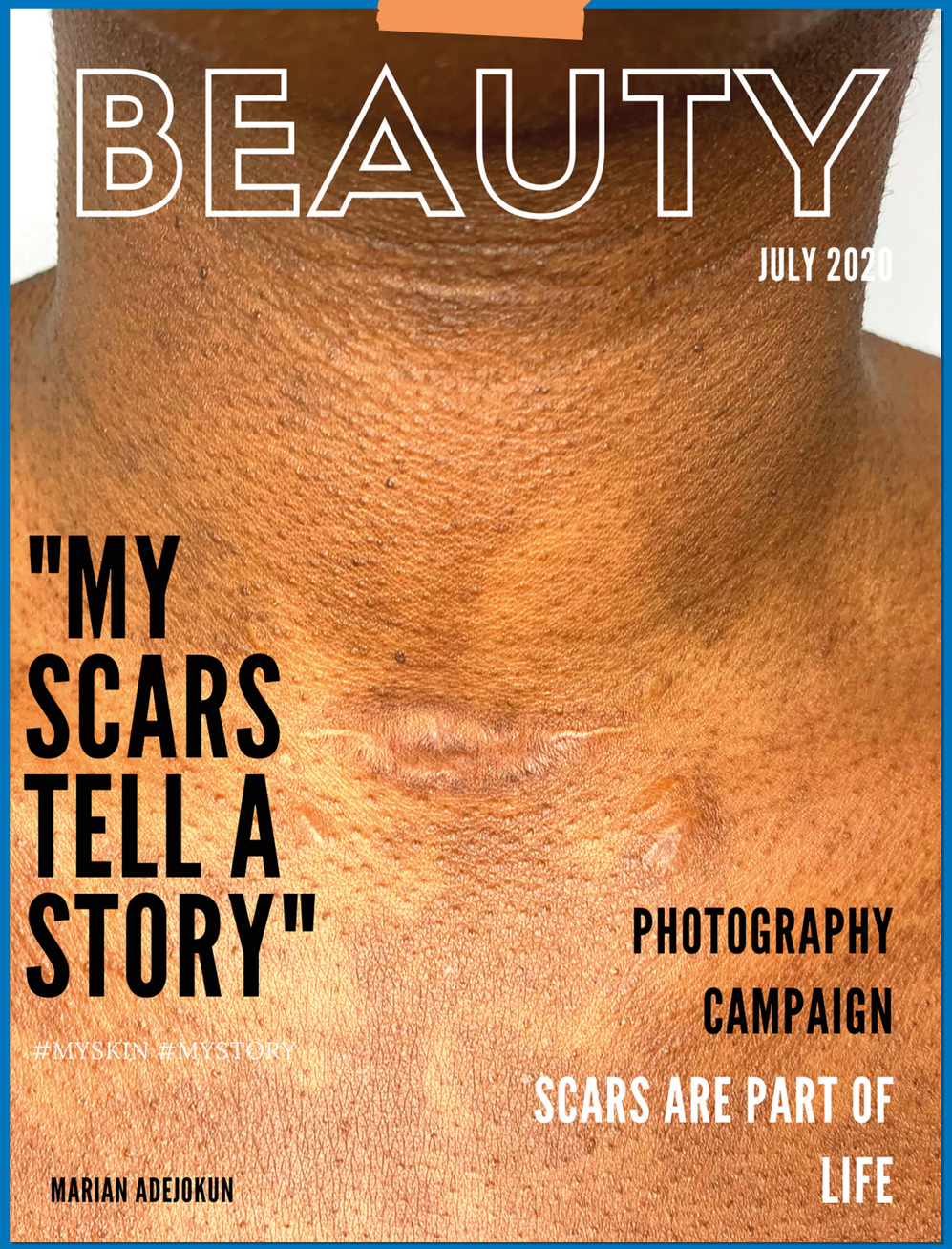
Skullduggery started a long time ago
With cunning that leads to perdition,
But Reynard's the fox who can still steal the show.

Nailbed capillaries magnified glow
With signs that persuade your clinician:
Three cheers for the Frenchman called Maurice Raynaud.

Animal stories continue to flow
With characters full of ambition,
But Reynard's the fox who can still steal the show.

Pills and infusions can blessings bestow
And reading provide some remission:
Three cheers for the Frenchman called Maurice Raynaud –
But foxy old Reynard can still steal his show!

*('Raynaud' rhymes with 'Say no')



My scars tell a story

Marian Adejokun, Croydon
Stevens-Johnson syndrome

My Scars Tell A Story Photography Campaign focuses on medical representation of diverse skin tones (diverse ethnicities) in the sector

- Medical presentation for students and professionals
- Awareness of SJS
- Promote body confidence & self esteem
- Break stereotypes in the media & social media

#MyScars #MyStory #MySkin

Scars of sound

Evie West, Redditch
Wolfram syndrome
Submitted by Wolfram Syndrome UK

'Evie entered the Y9-Y11 Worcestershire Poet Laureate with the poem "scars of sound". The poem was so incredible it won first place.

It's not just poems and English literature Evie is proud of, she was also trialling for the Women's deaf football team. Evie is a keen sportswoman and loves to play football and attend football games with her dad.

Evie is currently studying for her A levels in the hope of attending university to study architecture. Being deaf has not been a barrier to Evie achieving success in her chosen subjects.'

They are a reddish hue in colour.
Faint but visible to the naked eye,
weaved and intercrossed with thread
a day I so fearfully anticipated with dread.
They do not make me shed a tear. Nor cry
but are instead a reminiscence of my story.
The battle I fought to hear again;
they are my scars, my scars in all their glory.
My scars of sound. Till' the end

of
  forevermore

my spirit dances with them;
lifts up my heart. And soars.

To be gifted my magic ears,
       was worth the scars -
Almost ethereal. A planet like Mars.
Spinning, spinning. Vertigo is winning -
My head laced in white, plasters shall be off tonight -
oh' my scars.
My scars of sound.
The beauty fills you,
       with Earth's most utmost splendour,
Never fails to astound.
The queen of scars is crowned.

Have courage, I was told.
By all those, young and old -
But how. Could they ever know?
How much it terrified me,
to think
I could live

       or go.

I wear my scars with dignity, complete pride;
They make me complete; laced on either side;
a part of me that shall never forsake;
they remain, with all their hurts and aches,
a momentary flashback;
the injection of the cannula;
barely audible, a grunt of pain.
Outside, God sent down his rains.
Pitter
      patter.
Darkness.

 Sudden light.

My scars show my fight.
And I'll love them
    - for all my life.

Fight like a CHAMP1ON

Stacey McPherson, Stirling
CHAMP1

This isn't the life we expected,
This isn't the life we had planned,
Many a time we've been knocked down,
But we rise up again, here we stand.

Into the world came a gorgeous baby boy,
Our hearts and lives instantly filled with so much joy.

Ten fingers, ten toes, so precious our son,
No one could have prepared us for what was to come.

The milestones were missed and then the worry came,
Our life as we knew it was never to be the same.

You were different from others, not walking or talking,
You signed and you shouted, boy there was no stopping.

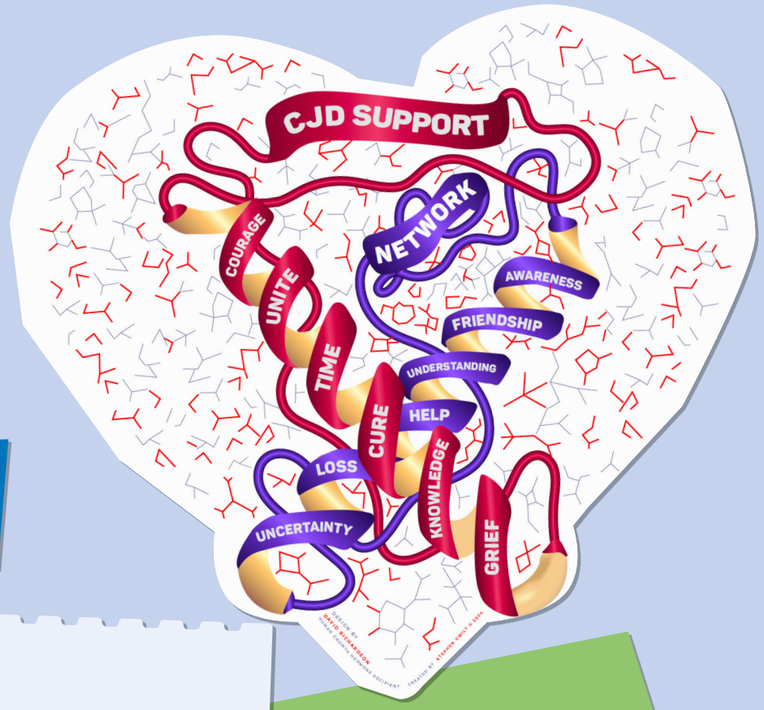
The meltdowns in public all the people would stare,
Looking back now why did I care?

Years of wishing and hoping,
Then grieving what's gone,
We didn't know then that the best was yet to come.

We finally got answers – CHAMP1 we were told,
Now we know what made you act so bold.

Your diagnosis is so rare so much is unknown,
But we will continue the fight – we will carry on.

To those who show pity, you prove them all wrong!
Don't pity this boy, he made us so strong.



Deep in the prions

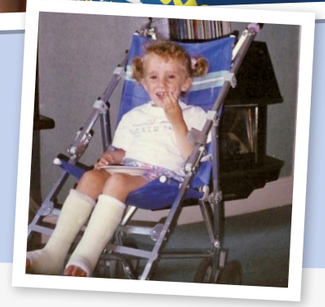
Dave Richardson, Yorkshire
Prion diseases, including CJD
Submitted by Beth Marsh

'As a young person, Dave Richardson received human growth hormone injections which were potentially infected with the rare prion disease CJD. As someone who is at increased risk of CJD, Dave is an active member of the UK prion disease community, raising funds and sharing his experiences to promote awareness and support others. Dave's original drawing, a creative interpretation of the PrPc protein (which misfolds into the disease causing prion protein) which depicts words representing the experience of the prion community, was created into a graphic format by his brother-in-law Stephen Gwillt.'

But you look so normal?

Charlotte Proud, Gateshead
Osteogenesis imperfecta type 1 brittle bone disease

'This was a poem that I wrote about my condition, Osteogenesis imperfecta, that is named after a consultation that I had with an Orthopaedic Doctor a few years ago. When I was younger, I hated having my condition, but now I love to share my story and hopefully inspire others. I feel as though my poem explains the difficulty of living with a rare disease that has no cure and is invisible. It also explains how determined I am including how I love exercising, particularly running and studying Mental Health Nursing.'



But you look so normal, how can that be?
Rather intriguing Orthopaedic Doctor once asked me.
In the room, myself, my Dad and another Doctor stifled a laugh.
This was a memorable appointment, and definitely a great gaffe.
The rare disease that I have causes my bones to be very brittle.
Latin is it's name; my first fracture occurred when I was only very little.
Easily hidden and invisible to others that I may walk or run past.

Broken bones, I've lost count, and many that have ended in a plaster cast.
Osteogenesis Imperfecta is rare but I am proud and I do love to tell others.
Never sitting still; cycling, running or playing football with my brothers.
Eager to always challenge myself and a medal to finish is always great

Determined I am; 4 marathons and 15 half marathons completed to date.
I am studying Mental Health Nursing and this is actually my 2nd degree.
Sunderland is where I am based at, such a brilliant supportive University.
Every hospital visit, ambulance trip, fracture, or surgery can be tricky.
Although I'm always grateful for the amazing treatment and staff that I see.
So although my rare disease is unpredictable, genetic and has no cure.
Everyone knows that I'll never give up, that's for sure!

Living with a rare disease without a cure,

is most definitely more life changing than you can imagine

Nicola Whitehill, Southport
Systemic sclerosis (SSc scleroderma) and Raynaud's

In 1997 aged 24, my life as I had known it, was to become no more.

I was told that I had a rare disease, to which there is no cure, little did I know, as to what was about to lie in store.

My symptoms of swollen stiff fingers along with very tight skin, difficulty with swallowing, all prompted diagnostic tests to begin.

A specialised blood test, confirmed diffuse systemic sclerosis, also known as scleroderma, along with Raynaud's phenomenon, with little medical understanding of cause and cure, at best.

A life expectancy of fifteen months was predicted, combined with immunosuppressant and steroids as a temporary fixative.

The following year, I transferred my medical care to the world expert doctors at the Royal Free hospital in London, and to this expert centre I attribute, my still being here.

Those initial years were touch and go, with frequent chemotherapy drips, back and forth to the hospital, like a yo yo.

Thankfully my name was removed from the stem cell transplant list, but by now I was unable to make a fist.

My body had become the real life tin man, and my life into the world of disability began.

Years of immunosuppressant, chemotherapy cocktails were my norm, in an attempt to calm my autoimmune storm.

On 1st March 2004, I achieved my dream of becoming a barrister, and, after 7 years of chemotherapy, I said no more.

Unfortunately, the disease has caused irreversible damage, making it a full time job to symptom manage.

Prevention of a Raynaud's episode is key, preventing gangrene, antibiotics and amputation, ultimately.

Living with a degenerative rare disease without a cure, is character building for sure.

Medical research is my only hope to improve what lies ahead, and, I actually do most of my #SclerodermaFreeWorld awareness hobby, whilst in my bed.

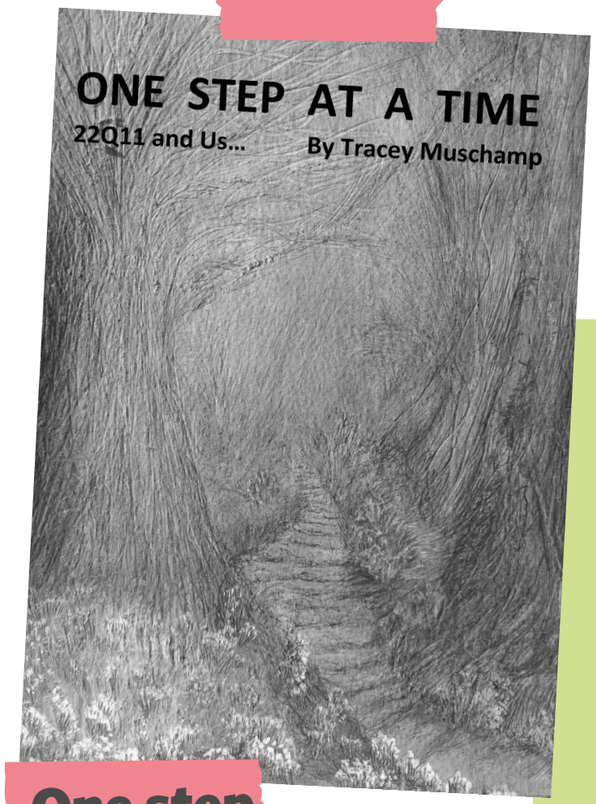
And although I am grateful to be 'semi fixed', each day is a relentless challenge, with new symptoms added to the mix.

Rare disease patients must unite, as we all have several commonalities within our plight.

Early diagnosis, medical expert centres, access to innovative medicines and medical research, to highlight just a few, to the rare disease patient, these are nothing new.

I have now spent over half of my life living with this body and dream hijacker, with the best medical team and hope as my elixir.

Living with a degenerative rare disease without a cure, is most definitely more life changing than you can imagine.



One step at a time

Tracey Muschamp, Scarborough
DiGeorge syndrome, velo cardio facil syndrome 22q

'We have velocardiofacial syndrome 22q/DiGeorge syndrome and have over 180 different problems from physical to mental, speech to hearing, we're all under a lot of specialists too and we also have to travel to different hospitals in Yorkshire.'

Your journey, more than we could imagine

Aileen Burnett, Blair Drummand
Undiagnosed genetic condition, hydrocephalus, autism, learning disability, ADHD, CVI

'He's writing the book', is what I say
We don't know what will be written day by day.
Now so many chapters full of joy and success
Though you do cause us, just a little stress.
Unwritten chapters waiting to be written
And still in the dark as to what's this condition

Your path unwinds, you go with the flow,
With each step forward, your story grows.
The meltdowns, the battles, the fears, the pain,
Hospitals, therapy, surgeries you sustain
Appointments, appointments, what's on today?
You're just a wee boy wanting to play

In a world that often tries to fit us in,
You really are a puzzle that refuses to give in.
You're a reminder, we're all meant to be different
You prove being different, really is magnificent.
Teaching us all as we watch you grow
What's important in life is not what we thought so.

You're writing your book, paving your path
And never, ever, looking back
Your hearty laughter, a joy beyond all bounds,
A big bear cuddle that lights faces around.
With many a chapter yet to come,
there's one thing's for certain, you'll shine as our son.

Life with you and all that you teach us
A journey much more than we
could have imagined for us.

I love you more

Lara Baxter, London
Progressive supranuclear palsy

‘My mum was diagnosed with progressive supranuclear palsy (PSP) in 2011. It is a rare neurological disorder which eventually stops a person’s ability to move, eat, speak, blink or see. She passed away in March 2022.

I wanted to write a poem that fought back against the pain and struggle of this powerful disease in the same way she did. Although it was all-consuming, I am adamant PSP and the sadness of those final, difficult years of her life should not be the lasting feeling of what our relationship was — it should be about strength and love.’

I love you more than the proteins
in your brain love to run riot.
I love you more than your truant nerves
could know, or your stubborn right foot
dragging along the floor.

I love you more than my temper boiling
because you tried again to walk,
face frozen and eyes wide, as surprised as me
to see you fall backwards like glass.

More, I love you more than your will
to eat, wash, function alone, a mind of pride
in a body of straw, still, I love you even more
than your heart’s hunger to stay beating.

And even though there are more
doctors with faces of questions
than understanding, more
healthcare cogs refusing to turn, more
doors opening shutting, jigsaws of district
nurse social worker unanswered
ringing phone lines—

I will love you more
than your spirit loved life,
loved choosing curtains and raising her child,
satin shoes and smoking,
LK Bennett and lamb shawarmas,
beautiful places among the palm trees.

Much more than the largest sky falling in
on a girl of nineteen unable to quite say it
then to the mighty woman I hope knew love.

Good striations

Helen Kurtz, Hevingham
Immune-mediated necrotising myopathy

‘I designed this blanket after being inspired by images of healthy muscle fibres. People who have idiopathic inflammatory myopathies (myositis) don’t have healthy muscle fibres. The bundles of striated fibres are represented in waves of knitting. This blanket was designed to help raise awareness of myositis and to fundraise for research.’

More than Leukodystrophy

Aisling Finn, Dorridge
Leukodystrophy - unknown type

‘I wrote this about my son. Originally we were told he wouldn’t live beyond two, that changed to they didn’t know. Yet, we are at seven and he is more than his diagnosis. We do not know what the future holds but we know he surprises most who meet him after reading his notes.’

He is more than the word, ‘Leukodystrophy’
He loves to keep colouring within the lines,
but he lives his life outside a closed
thinking box that came with his diagnosis.
For he can do all the things they
suggested would not be possible
He can hop on one leg, run, climb
- boy can he climb!
He can problem solve
He won’t be told, ‘not today’
He’ll have a go before he takes a rest
For he is more than Leukodystrophy
He is seven, living his life
Learning about his challenges
But not taking ‘no’ for an answer.

Caring down the cracks

More layers than you can imagine

Jillian Shields, Glasgow
Suspected mast cell activation syndrome

‘This poem was written based on the theme of Rare Disease Day 2023 which was Care Coordination. The poem was inspired by thinking about this theme and what it could mean. Revisiting it in 2025, I think it speaks to ‘more than you can imagine’. Caring for someone with a rare disease involves more layers than you can imagine.’

I’m Rare,
Or maybe not
But I seem to be
When it comes to Care.

All the labels
Don’t fit me
I’m in between two
Or a combination of three

Every consultant I see
Has their own window view
But I’m a prism
They can’t see through

My symptoms
Are many colours
Only some
Are the same as others

It’s all in how they bend
And refract
Some disappear
And retract

To see in full
Takes a step back
So you can see down
Through the cracks

So, consultants, wrap around me
Like orange peel
And your combined efforts
Will help me heal.

More side effects than you can imagine

Mellissa Monsoon, Ramsgate
Idiopathic intracranial hypertension (IIH)

'When diagnosed with IIH, you are given medications which are used to decrease the spinal fluid in your body, reducing the number of migraines. However, the side effects that come with these medications are often not communicated by GPs, so patients often confuse the side effects of the medication to be symptoms of IIH.'

It will not grind us down

Phillippa Farrant, England
Duchenne muscular dystrophy

'I am one of Genetic Alliance UK's trustees and about 2 years ago my band wrote a song about the fight with services for all the work I had done fighting to get things for my son who had Duchenne muscular dystrophy.'

The system tries to snare us in
To pull us down and make life hard
Our boys decline as time goes on
IT WILL NOT GRIND US DOWN

We fight for this we fight for that
The schools don't help they need more funds
The NHS... what NHS?????
IT WILL NOT GRIND US DOWN

MPs come and MPs go
They promise this they promise that
Our boys get worse... whose fault but theirs?
IT WILL NOT GRIND US DOWN

They change the rules
More this less that
No social care. No carers pay
Keep struggling on is all they say
IT WILL NOT GRIND US DOWN

Equipment, wheelchairs, carers more
We fight our corner still and more
Commissioners sit and smile at us
IT WILL NOT GRIND US DOWN

We love our boys we want what's best
We know their needs we KNOW what's best
The cough assist, the NIV
IT WILL NOT GRIND US DOWN

And as our boys decline in health
We rant and rave, we grow so old,
We fight their fights, we push their needs
IT WILL NOT GRIND US DOWN

As time goes on, we need still more
We write the rules, we break the rules,
We fight the world, we lead the world,
IT WILL NOT GRIND US DOWN

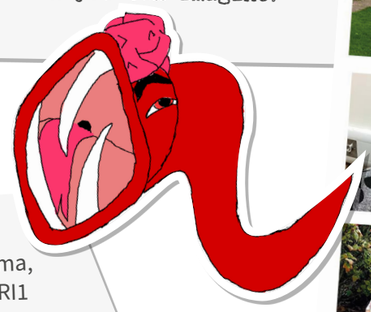
So angry mums and angry dads,
We fight and pledge. Till we have won
The system stinks... but you know what

IT WILL NOT GRIND US DOWN!

More than you can imagine

Hannah-Louise Blackall
Thrombotic thrombocytopenic purpura (TTP)

'What's that?' people have said
more than you can imagine.
Dentists back away.
Doctors furrow their brows.
Nurses look concerned.
And that's just rare condition number one which I have had forever.
A full life, a surprise for many
Then ultra rare number two arrives to stay.
More times than you can imagine I have seen glazed looks of 'Is she bonkers?'
Or the narrowing of the eyes: 'Did we cover this in med school?'
Or, 'Oh God please let her not be my patient!'
But those in the know are different.
They smile and exude calm,
battle plan already in hand
The hidden danger to them a familiar
foe.
New weapons now
advice not to google is sound.
The lethal sting off TTP less deadly now.
Not long ago a different story.
For Eli Moschcowitz and all of you
who have moved the story on
Gratitude more than you can imagine.



Kat Miller, Ayr
cerebral cavernoma,
KRIT1 type, CHIARI1

'I was diagnosed with cavernomas at 8 years of age, currently I volunteer for Cavernoma Alliance and create stories and characters related to my journey with cavernomas. This piece symbolises one of my Cerebral Cavernomas when it is triggered, the thrashing and red angry look portraying the intensity of the pain that they cause.'

Markus Bell, London
Neurofibromatosis Type 1 / Type 2 – related schwannomatosis and schwannomatosis

'Photo collage of the nurses involved in the charity Nerve Tumours UK, making the everyday better! Having access to a specialist NF nurse remains a postcode lottery – we are growing geographically and our helpline has just the same function if your region is not covered.'

PNPT11 Noonans I screamed

Hannah Doyle, Halifax
Noonan syndrome

'A poem about my son who was initially found to have a 5q15 deletion and also Noonan's that was missed pre-heart surgery resulting in a life limiting catastrophic global brain injury.'

As soon as he was born it was clear
He's just a typical baby my dear
But really he's not I pleaded
All the observations they planted a seed

Ok we'll take some blood
I fully understood
Up to 3 years they said
3 years if serious he could be dead

He needed open heart surgery
Surgery, oh no first came the pre surgery
He's got Noonan's I kept saying
We need to proceed so I was just praying

All seemed good
But he had Noonan's
why could they of understood
A nightmare struck
This wasn't in the book

Never the same
We had no idea about all that came
He had Noonan's too I screamed
Noonan's and a catastrophic brain injury
they screamed.

The pain is the worst

Jennifer Jones

Poem created from discussions with researchers funded by Scleroderma Clinicals Trials Consortium.

The pain is the worst
You want to sit there and rock,
the pain is the worst
as if you've taken the skin off with a cheese grater,
the pain is the worst
like rose thorns stuck in your finger,
the pain is the worst
as though you've scalded your finger,
the pain is the worst
like I just slammed the car door on it,
the pain is the worst.

Your finger is in the hinge of the door,
the pain is the worst
hammering a nail right through the tip,
the pain is the worst
like somebody's sticking a needle in your finger,
the pain is the worst
you just rock back and forward,
the pain is the worst
just want to sit on the floor and cry,
the pain is the worst.

You hit them,
they hurt
and to actually bend the fingers,
they hurt
where your ulcers are,
they hurt
actually on top of the knuckles,
they hurt
the pain never goes,
they hurt.

The pain is just unbearable,
chop your finger off
you want to bang your head to refer the pain,
chop your finger off
it's just so painful,
chop your finger off
just to release the pressure,
chop your finger off
that's how bad it is, the pulsating pain,
chop your finger off.

More puzzling than you can imagine

Pamela Coombes, Cardiff
Barraquer-Simons acquired partial lipodystrophy syndrome

Invisible battles

Rhyanna Halasovski of Studio RLH
Pregnancy associated osteoporosis (PAO)
Submitted by Karen Whitehead MBE, Somerset

'This artwork was produced by Rhyanna while participating in a Translating Science Through Art project and it's dedicated to the rare disease community and the resilience of patients and their families, who embody hope and progress in every endeavour.

The work delves into the profound emotional landscape of rare disease isolation and the severed bonds that accompany it. It evocatively captures the patient voice by showing the experiences of women with Pregnancy-Associated Osteoporosis. 'Invisible Battles' captures the silent struggle of the unseen, the misunderstood. It embodies the universal battle for recognition and understanding in the face of undiagnosed or rare diseases. It also eloquently conveys the isolation and vulnerability and overwhelming nature of coming to terms of having a rare disease diagnosis, while additionally hinting at a long rare disease journey ahead.'

Thea: more than we could ever imagine

Clare Astle, St Austell
DYNC1H1 genetic mutation

'This has been written to summarise the journey we, as parents of a child with a rare genetic mutation, have been on since her birth – how wonderful it has been but how emotional and hard it can be too.'

She grew. She flew.
She is so much more than we could ever imagine.

Waiting for so long; we never could imagine
Never did we think she would be anything but 'normal'
That word we've come to know
Does
Not
Exist.
Born. Beautiful. Everything we imagined.

Days, months passed and she grew
But not like the others
Not like those who smiled
and laughed and cooed and rolled and sat and stood and walked
Reassurance came: they all develop at their own pace
Deep down we knew
Something
Wasn't
Right.

Diagnosis followed a test 'just to check'
Relief
Uncertainty
What now?

What now? She grew! She flew!
She smiled and laughed and cooed and rolled and sat and stood and walked.
No words are spoken but she lets us know just what she wants.
She spells with magnets. And reads them too. She signs in her own way.
She learns to swim.
She charms
She loves.

Our rare beauty makes us more than we were before
She's made us better people
Hard days, hard nights
Life isn't easy
It's all unknown, it's all uncertain, it's all an adventure with her.
It's all so much more than we could ever imagine.

She grew. She flew.
She is so much more than we could ever imagine.

I have known too many ambulances

Anonymous
GRIN1 gene disorder

One year we had seven trips; three with blue lights.

The first, when something went very wrong in Tesco. ‘Take some video’ the paediatrician had told me, ‘if he does anything unusual’. I filmed for 15 seconds until scientific curiosity was overwhelmed by mothering instinct and I let out a yelp for help… and a passing fire crew were quick to corral some trolleys around us where we lay, on the shop floor, to shield us from prying eyes and other wheels until the cavalry arrived.

The middle ones are a bit of a blur. We got good at filling the gap between calling 999 and departing the house with bag-packing, not knowing how long we’d be away but squeezing in a few extra nappies for him, and spare undies for me. Mostly, it was only one night.

The sixth, I remember well. An autumnal cafe, a cold clear day, a cobbled courtyard. Left the boy in his buggy at a sunny table, to join the coffee and cake queue. Came back and he’d stopped breathing. Girl Guide First Aid kicked in and I slapped him between the shoulder blades until his airway cleared. But the registrar wouldn’t let me take him home again without a refresher CPR tutorial, just in case…

The seventh, even the paramedic told me to hurry up with the packing and jump into the van. There was a whole crew waiting in resus for us, a semi-circle of aprons, badges and grim faces poised around the blank space where the stretcher swept in. Someone offered me a plastic chair, but sitting down was the last thing I wanted to do. Nameless people leaned over him, ventilated him, injected him while I peered on from the edge.

Bonus extra trip: the seventh didn’t bring him back from his seizure so we got the VIP treatment, to a different hospital with intensive care. ‘Do you get carsick?’ they asked. ‘Because we go really fast. And we’ll only stop if we need to work on him’. Free Mars bar and overnight toiletries from this gang; dead fancy. Absurdly comforting in the centre of a tornado.

After that, they concluded he had epilepsy (among his other issues), and we started on medication.

The next years were quieter, but we still needed to stay sharp. One trip, we left the wheelchair behind and had nowhere safe for him to wait at A&E, except on my lap, for several hours. Once, his dad had to run down a mountain to join him in hospital. Twice, I was out of the country with work. Neighbours leaned in.

One blue-lighter, I got to look out the front. I saw a mighty parting of the Red Sea – the kindly drivers of Bristol pulling over to let us through. Of all the memories, that’s the one which still makes me cry.

More waiting rooms than you can imagine

Aisha Seedat, Leicester
Morquio A syndrome

‘The reflection describes the experience of someone with a rare disease who spends a lot of time in hospitals. They attend numerous appointments, undergo tests, and try to find treatments, but often feel stuck in a cycle of waiting. The waiting rooms represent the uncertainty and isolation they face – waiting for answers, results, or relief that never seem to come quickly enough.

Living with a rare disease often means that doctors may not have all the answers, and treatment options can be limited or unclear. The long hours spent in these rooms feel endless, and it’s easy to feel like life is on hold.’

It feels like my life is measured in waiting rooms. Endless chairs, sterile walls, ticking clocks that never seem to move. Some days, I forget what I’m even waiting for. Another test, another specialist, another round of questions. They ask about my pain, my progress, my fears, but it’s the silence in between those questions that feels the heaviest.

I’ve spent years in these rooms, each one a reminder of how much of my life is out of my control. I’ve met strangers in white coats who offer hope, but most of it feels like a promise I’m not sure I’ll ever see. More waiting rooms than you can imagine. Each one holds pieces of my life, fragmented and uncertain. But as I sit there, I remind myself — this is part of the fight. I’m here. I’m still fighting. And somehow, that’s enough.

More impactful than you can imagine

Anonymous, Preston
Acquired nystagmus
Submitted by Jackie Roberts

Eyes bleary and frantic, world spins,
Unbalanced, nausea, fatigue,
‘Wobbly’ vision, ‘drunken’ gait,
Calm yourself...

Family member/friend is driver and helper,
Too much traffic, too little parking.
Busy corridors, harsh lighting,
Signs ‘move’, so can’t be read.
Visual stress! Body and mind in perpetual flux.
Calm yourself...

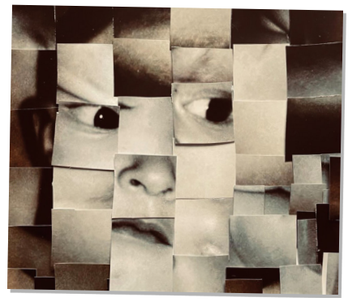
A long, hot wait – my name is called,
A weary smile, head down, reading.
A knock at the door – more notes brought in.
Not mine. Other people’s lives intruding,
impinging. The clock ticks silently.
My ‘mask of capability’ hosts an unconscious
tug of war, with my new, vulnerable self,
Kindness and patience will tease out the latter.
Cursory checks, sometimes more thorough.
Hopes deludedly high, expectations often low,
Since, there’s ‘no cure.’

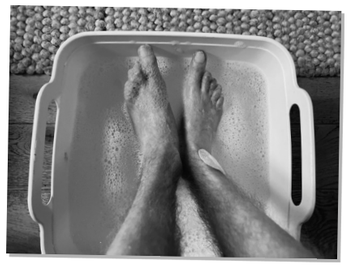
Symptoms worse; more cross-over drugs to try,
Side-effects are no joke and no lure.
This is ‘your condition to manage.’
Heal yourself...

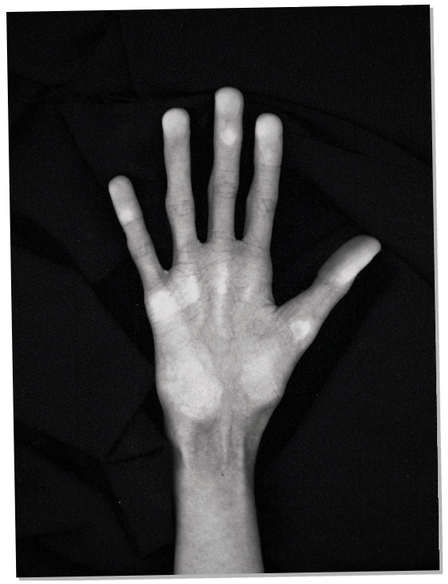
More extraordinary

Dylan Lombard, Glasgow
MDP syndrome - mandibular dysplasia with deafness and projeriod features

'This series of photographs was about exploring the beauty of my body and my own self image. The photographs show the uniqueness of my body and show that it's beautiful to me. I sometimes feel like I am forced to hide my arms or legs because I feel ashamed of what people might say but my passion for photography has helped me become more confident in my body.'



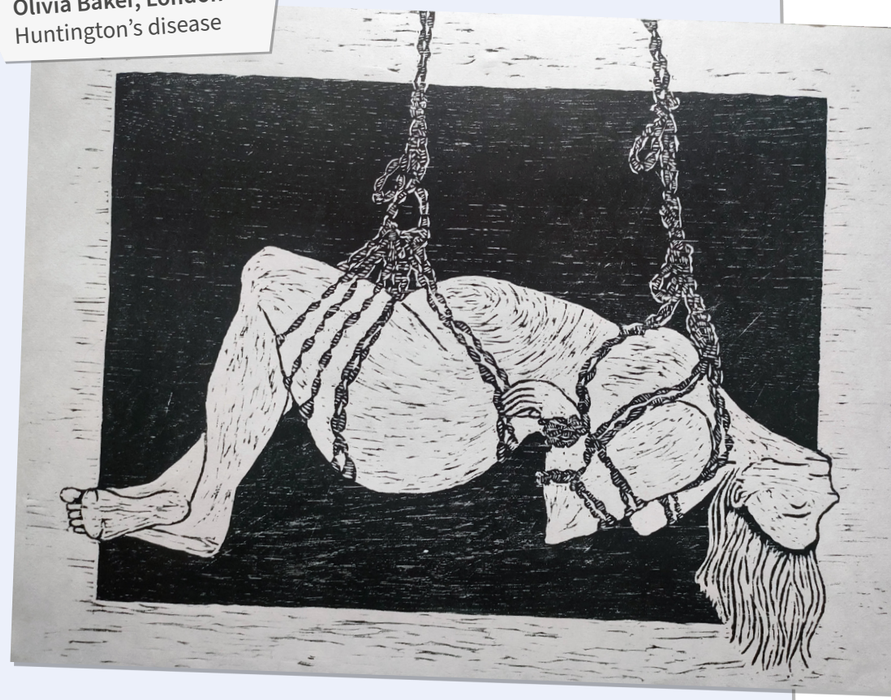




50/50

Olivia Baker, London
Huntington's disease

'Huntington's disease is an autosomal dominant condition, it comes with a 50% risk of inheritance, a 50% risk of passing it on. Even when you find out your result, there is nothing that can be done, you have to just hang on and wait for your DNA to change who you are. When your baby bump comes back gene positive, you have to pick what the humane and loving option is. It might be 50% inheritance, but for everyone involved it is 100% heartbreak.'



To my brave heart

Bradley Sinfield, Peterborough
Spinal muscular atrophy

In the quiet strength of morning light,
Where shadows yield to courage pure and bright,
There stands a spirit, fierce and gently true,
A heart that faces trials with a view.

Though life may weave its path with threads so fine,
And challenge you with storms that intertwine,
Your bravery shines like the morning sun,
A beacon guiding all who see you run.

With every step you take, a story told,
Of strength and grace and heart that's brave and bold,
You turn each trial into paths anew,
A testament to all the things you do.

In moments hard, where others might despair,
You rise with grace, a strength beyond compare,
Your spirit dances in the face of pain,
A melody of hope amidst the rain.

So let the world behold your shining light,
A courage that turns darkness into bright,
For in your eyes, a strength so deep and clear,
A reminder that the bravest hearts are here.

More support than you can imagine

Omi Gates, Leominster
Salivary duct carcinoma

'It's frightening to be diagnosed with a rare and aggressive cancer, so I wanted to create a piece of art that shows how much I appreciate my people. My family and friends, my fantastic medical team, my fellow warriors, and Emma and the team at Salivary Gland Cancer UK; a charity who work hard to raise awareness, and funds for trials and research to find better treatments and cures for these rare salivary gland cancers, and who offer priceless support for people like me.'

See me

Kay Mullan and Eli,
Cookstown, Northern Ireland
Rare chromosome disorder

I may have lost my ability to speak but I can communicate by body language and through gestures.

'This is little Eli who has a unique rare chromosome disorder that brings many struggles for him every day. He lost his ability to speak when he was just one. He is non-speaking, with a severe learning disability.

This is a very emotional, caring and respectful post capturing Eli's expressive eyes and hands making simple gestures to convey emotions and thoughts. He is surrounded by a comforting, warm aura, highlighting his resilience and the support he receives everyday by those who love him.

The mood is bittersweet, with a focus on his true bravery and his silent but powerful way of expressing himself.'

#SeeMe raising awareness for all non-speaking people.

My journey - to Hell and back

Rosie Novis, Billericay
Guillain-Barré syndrome

'I was initially diagnosed in November 2021 with Guillain-Barré syndrome but with a ? In early 2022, the diagnosis was upped to Chronic Inflammatory Demyelinating Polyneuropathy. In November 2024, following further investigation, it was confirmed that I had Guillain-Barré syndrome.'

Three years lost to a rare and cruel condition,
Guillain-Barré syndrome,
Incredible ICU unit, peaceful and amazing care,
Tracheotomy and machines, often feel very hot,
Legs numb, bedridden, unable to speak or swallow,
Persistent speech therapists, helped me find my voice again.

Occasionally angels' wings touched me –
caring, bringing love
Skilled nurses and doctors took every care of me,
Chaplain visited every day, bringing hope.

Move to a general ward, isolated in single room,
Long, long, lonely days,
No neurologist or special nurses to talk to,
Waiting and waiting for someone to help or to explain,
Night times full of dreadful, frightening dreams,
Traumatic staff incidents engraved on my mind.

Occasionally angels' wings touched me –
caring, bringing love
A sister helps me shower after her shift ended.

Move to a rehab ward at a small local hospital
Over a week's wait for the rehab unit
Long, long, lonely days
Lying in my bed, unable to do anything
Nobody to talk to or ask questions
No reassurance

Occasionally angels' wings touched me –
caring, bringing love
Another nurse gave me a gentle chair bath,
washed my feet.

Home at last for Christmas,
Tears of joy, Christmas lights twinkling,
Welcome home, safe and loved.
A fall takes me back to hospital,
CIDP diagnosed, with immunoglobulin infusions,
Eighteen months of tedious infusions,
collapsed veins,
And long days, long ambulance rides.

Occasionally angels' wings touched me –
caring, bringing love
A young nurse, remembers me from pre-ICU,
and finds time to talk.

Back in hospital with anaemia, huge bruise on leg
Blood transfusions, tests again
Nobody tells me why, or what caused it all.
A & E consultant tells me I have blood cancer
May only have two days to live.
But nonsense – too weak to make a complaint.

Occasionally angels' wings touched me –
caring, bringing love
Witness a porter comforting a frightened old lady,
He takes her for an X-ray

Request referral to National Hospital for Neurology & Neurosurgery
Wonderful care again, all medics and nurses
Who speak to you as if you are a real person, not a lump in a bed
Tests and more tests, waiting for results
Results all clear, all clear, I have not had CIDP!
But still remain with GBS, no more immunoglobulin.
Unable to walk far, or to go out freely.
I am well. All I need is a new pair of legs!

Occasionally angels' wings touched me –
caring, bringing love.
I learn about acceptance, keeping strong,
not giving up.

Thankful that I have survived, grateful to be alive
My beloved husband cares for me uncomplainingly.
We are together – each day is special.
Most days angels' wings touch me –
caring, bringing love.

More complex than you can imagine

Danielle Alexander, Scotland
Mitochondrial disease

I had a complex and challenging medical history where I was described as being a 'problem patient', physical and psychological causes were explored but there were no answers.

My diagnostic journey has, felt like a never-ending rollercoaster.

My life felt like a tug of war between medical and mental health professionals.

I felt trapped in the wrong service, like a criminal who had been wrongly convicted.

Two years ago, after 33 years of medical mayhem, I received a diagnosis. A condition I had never heard of until this point, Mitochondrial Disease.

100,000 genomes project finally gave me the answers I had been looking for.

My geneticist once said that finding an answer would be like trying to find a needle in a haystack.

I have always been determined, I refused to give up until that needle was found.

As some people are head-hunted for a job, in my case I was head-hunted for a diagnosis.

My life and many others could be drastically different and potentially improved by a faster, accurate diagnosis.

Mitochondrial disease is progressive and can be life-limiting.

My mission in life is to do all I can to help raise awareness for mito - but also bring hope and empowerment for anyone still experiencing a lack of diagnosis and a medical wilderness.

I hope that one day there will be a cure or treatment for Mitochondrial Disease.

More lonely than you can imagine

Anonymous, Preston
Acquired nystagmus
Submitted by Jackie Roberts

Playing the clown when others are there,
Trying to fit in, so they don't stop and stare,
Not being the person to whine or to moan,
Letting the mask slip when you are alone.

Life was so easy before things went wrong,
I'd planned on my 'normal' life being quite long,
But fate intervened – I feel old way too soon,
Now I struggle to rise until closer to noon.

Others get on with their lives as before,
With energy, zest and a zeal to explore,
I try to join in the best that I can,
But more often than not I'm forced to replan.

At times just the most basic of tasks,
Remind me of limits where energy lacks,
I paint on a smile, but I don't know who for,
I think it's for you, so I won't be a bore.

But with the right people and in the right place,
I don't have to change what I 'say' on my face,
I'm open, I'm honest, I smile and feel true,
To the version of me that can be but not do.

More misunderstood than you can imagine

Eve Wisniewski
Type 1 HFE genetic haemochromatosis
Submitted by Haemochromatosis UK

'Haemochromatosis UK collaborated with Pulitzer Prize winning photojournalist Cathal McNaughton on an awareness project designed to showcase members' experiences with genetic haemochromatosis. Members of our community had their photo taken by McNaughton and their stories documented. As an 'invisible illness,' we wanted to depict the faces of genetic haemochromatosis and the stories behind them.'

George's story:

In 2021, the community group I run was approached by Haemochromatosis UK to collaborate on a community screening project here in the heart of the Creggan, Derry. We helped push the message out. We started off by testing our own team. When the results came back, I found out that I had haemochromatosis.

If it had not been for the community screening initiative, I probably would never have known that I had it. I would exercise regularly, and the nature of my work is pretty hectic, so I just put the brain fog down to stress and tiredness. I've noticed it drastically reduce since starting venesection treatment. I had a big rash on the side of my face and once I started getting venesected, it went.

I didn't know what to expect. It wasn't a significant diagnosis in my eyes, although once I looked into it, I thought, I'm glad I've picked up on this at the age I'm at rather than picking this up in 10, 15, 20 years' time.

"We started off by testing our own team. When the results came back, I found out that I had haemochromatosis."

"Whatever has been affecting me is ten years worse than it should've been."

Rita's story:

The doctor said, 'I don't know if I can send you for that test, I would get a slap on the wrist; it'll cost us a lot of money.'

They told me I had lupus. They said I have rheumatoid arthritis. The consultant scanned my shoulders and looked puzzled; she said, 'you have no sign of rheumatoid arthritis, have you heard of fibromyalgia?'

I sent away for a kit with Haemochromatosis UK and within about six days I got the test back and a letter that said, 'you are a carrier for haemochromatosis with the C282Y gene.'

I went, yes! I knew it! And the next minute I was in a flood of tears because I had to fight so hard to get it. I wrote a letter to the GP and said the test has confirmed I have the condition; I'm still waiting for a doctor to get back in touch with me. The doctors will no longer accept letters.

I started thinking, I'm ten years along the line of not having had this test so whatever has been affecting me is ten years worse than it should've been.

How do I feel now? Utter relief. Hopefulness. Gratitude. Emotional.

A rollercoaster experience - rarer than you can imagine

Kimberley Stewart Beasley, Glasgow
Dysfibrinogenemia

'Heavy bleeding during menstruation can be signs of a bleeding disorder in girls or women. Earlier recognition of girls or women with heavy menstrual bleeding and an underlying bleeding disorder is important to enhance quality of life. Menstruation is seldom discussed openly, I wanted to highlight the difficulties from a female perspective of living with a rare bleeding disorder. This was to help increase awareness and to highlight how difficult it can be to access care due to there being limited knowledge in healthcare about rare conditions.'

A woman stands in front of me

Looking as white as a sheet with pains as sharp as a knife

She is always at the mercy of what "Aunt Flo" decides

"Aunt Flo's" arrival this month brings a fast furious fountain overflowing with blood

Saturating her jeans, putting a spanner in the works and spoiling her plans to have any sort of fun.

This woman reflects me. Desperately, yearning for Dysfibrinogenemia to be understood!!

A bleeding disorder so rare!!!

Its impact always misunderstood!!!

I dread saying the "D" word over and over and again and again.

A stare always received like a deer caught in the headlights.

Fruitless conversations, hearing over and over and again and again, there is no information available online.

My heart sinks heavily, I wish I could put yourself in my shoes.

To highlight the day-to-day struggles of living with rare disease to you

How isolating would it be to feel alone and confined?

How continuously exhausting would it be to self-advocate and co-ordinate your own care too?

I hope to spread awareness of rare conditions to you and its challenges by writing this poem for you.

And reach out to those feeling isolated in the rare disease community too.

If health professionals were more rare-aware, there would be reduced isolation and effective co-ordination of care.

More... than you can imagine

Anonymous
GRIN1 gene disorder

'A few thoughts distilled from a decade of rare parenting and advocacy.'

More tiring *– the caring, the admin, the uncertainty never ends*

More challenging *– there's so much to learn, about people and systems*

More isolating *– a condition so rare that very few people have it, or 'get it'*

More frightening *– peers can suddenly die; is that our future, too?*

BUT ALSO

More passion *– 'fight or flight' turns into energy; harness it for good, for all*

More life *– small wins and inchstones can mean the world*

More authenticity *– see who people really are, and what really matters*

Simply: *more love.*

Living with a genetic condition, more complex than you can imagine

Joy Bowstead, Middlesbrough
Marfan syndrome

'A rose, so strong, yet so delicate.
Genetically interesting & complicated.
Studied by scientists. Admired by artists.
Loved by poets. Whilst varying in size and
shape, they inherit similar traits which
are passed down through their genes.'

More than you can imagine

Lucy Richards, Weymouth
22Q11.21 micro duplication

'Written about our son.'

L.R. sat cross-legged on the carpet, building a tower of blocks with meticulous care. Each piece had to fit just right, perfectly aligned, as if the whole world depended on it. His parents watched from the sofa, their hearts full of love and worry. L.R. had always been unique, living with 22Q11.21 microduplication, autism, ARFID (Avoidant/Restrictive Food Intake Disorder), global developmental delay, and GERD (Gastroesophageal Reflux Disease).

His journey wasn't easy. There were days when the smallest tasks felt insurmountable. A new texture in his food, an unexpected sound, or even a sudden change in his routine could set his world spinning. Doctor visits, therapy sessions, and special education plans filled his days. Each diagnosis came with challenges that stacked like the blocks he loved so much – precarious, teetering, and overwhelming.

Yet, L.R.'s world was not defined by his diagnoses. It was filled with wonder, creativity, and resilience that often left those around him speechless.

One afternoon, as sunlight streamed through the window, L.R.'s mom noticed something. His tower of blocks had transformed into a sprawling city, complete with bridges and tunnels. "L.R., this is amazing!" she said, crouching down to his level.

"L.R.'s world was not defined by his diagnoses. It was filled with wonder, creativity, and resilience that often left those around him speechless."

L.R.'s eyes lit up. "It's for you," he said softly, handing her a block painted with tiny stars. Mom felt tears prick her eyes. It wasn't just the city he'd built – it was the way he saw the world. Where others saw limitations, L.R. saw possibilities.

At school, L.R. struggled with communication, but his teacher discovered his talent for drawing. His sketches told stories more profound than words ever could. A shy classmate, inspired by L.R.'s art, began to open up, and soon, the two became inseparable.

L.R.'s challenges didn't disappear, but his family learned to see them as part of his extraordinary tapestry. ARFID made mealtime tricky, but it also taught them to celebrate small victories – a bite of a new food, a sip of water without fear. GERD brought discomfort, but L.R.'s determination to keep smiling through it amazed everyone around him.

One night, L.R.'s dad sat beside him as he stared out the window at the stars. "L.R.," he said, "you're teaching us so much – more than we ever imagined." L.R. turned to him, his eyes reflecting the starlight. "I just see things differently," he said simply.

And that was L.R.'s gift: the ability to show the world how to look at life through a lens of beauty, persistence, and creativity. His journey wasn't just about overcoming obstacles; it was about transforming them into stepping stones for something greater.

Because L.R. wasn't just living his life. He was building something extraordinary – more than anyone could have ever imagined.

It's awesome to be different

Luiz
Submitted by Janine

There is nothing wrong with being different
It would be boring if we were all the same
If we looked like each other
Or had the same first name.

Wide apart eyes,
Slanted nose
No arms or legs,
Or fingers or toes
It's ok to be different.

Some people are in a wheelchair
Or have eye-gazers to help them talk
A guide dog, to help people who can't see
And crutches to aid them to walk.
It's ok to be different.

Some have brains that are wired differently
And genes that are all mixed about
Others might not speak the same language
And a few just squeal, scream and shout
It's ok to be different.

Some people have a fantastic memory
Like me – they don't get bored
We are all unique in some great way
And want to be cared and adored
IT'S AWESOME TO BE DIFFERENT!

Rare

Amanda Cole, Tring, Hertfordshire
Malan syndrome

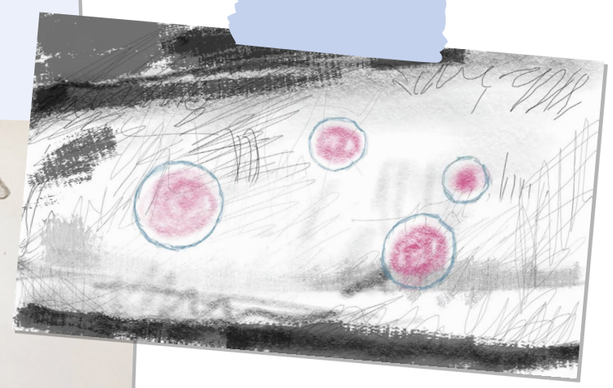
'All the pictures are of my son, apart from the top-right which is from a meeting of the Malan Syndrome Foundation. Bottom-right is my son and daughter.'

Living with Behçet's

Deborah Cardinal
Behçet's syndrome
Submitted by Behçet's UK

'In 2022, Behçet's UK members from across the UK were invited to participate in a creative project with Breathe Creative, an arts-based company from Wales, to produce an animated film on the lived experience of Behçet's Disease as a way of raising awareness on both the disease and the patient perspective.

The result is a raw, vulnerable and emotive film that for many patients, accurately reflects the nature of living with this rare, complex and lifelong condition.'



I've finally got some answers
but why am I so scared?
My future looks so dark and grey
It's like I'm lost and no ones there

The fear is overwhelming
looking out into the unknown
I've got so many questions
But the darkness is overgrown

There's relief deep down inside me,
like I'm finally free from chains
I feel like my life has vanished
But maybe I'll fly again?

Worth more than you can imagine

Will Newman, High Peak
Cystinosis

'This is dedicated to my amazing 9-year-old granddaughter, Ellie, who has cystinosis.'

The specialist looks up.
"It's rare, very rare.
I've never seen one like this before."

"How much is it worth?" I say.
A sharp intake of breath.
"It doesn't work that way,
Not like jewels or art or manuscripts.
This is ultra rare, you see."

"You mean that it's worth even more?"

"It's the crystals.
Every cell, every organ, every muscle,
every bone – Even in the eyes."

"That's amazing. So many crystals.
How many cells are there in the body?"

"About 30 trillion, give or take a few."

"Wow, that's a lot of shiny crystals, then,
Worth more than money can buy?"

My little cystinosis star.
Worth more than you can imagine,
Worth more than money can buy.

Through your eyes

Hope Miles, Cardiff
Long QT syndrome
Submitted by Tammy Miles

How can I see through your eyes
when your world is so different to mine?

I wish I could do it just for a day,
to see the world how you play

To know why you get sad and cry sometimes
To see why the world is so loud

You get angry and sad when you can't say out loud
the word that's trapped deep inside somehow

It's too bright

It's too loud

I can't see the world through your eyes

I don't understand, I wish I could

But I will always stand by your side

My brother

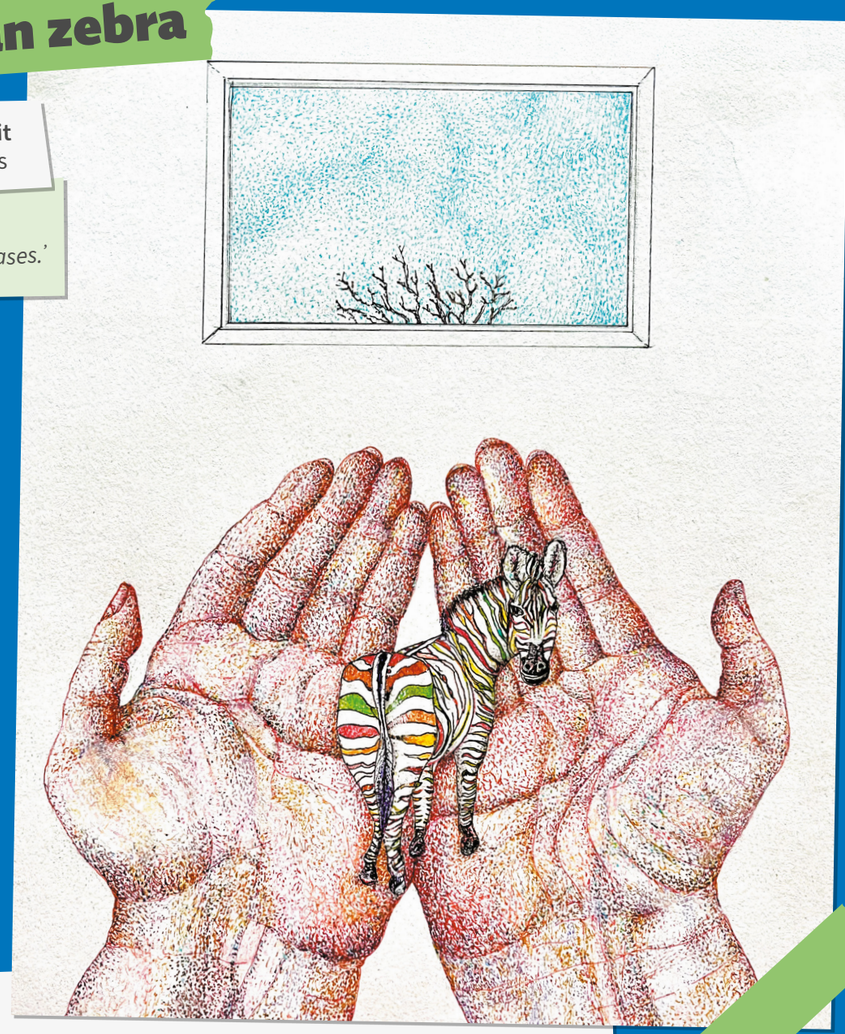
My twin

My everything.

More than zebra

Matilda Tumim, Wormit
Multiple rare conditions

'A poem for those with ultra orphan (rare) diseases.'



The tiny zebra trots along
Unaware that she doesn't belong
She is surrounded by her kind
Absent monochrome they don't mind

When hungry lions chase for snacks
They only see the black and white backs
Rainbow zebras are so rare
Beautiful beyond compare

A lotus flower and me... connected by creativity...

Lucy Alexandra Atkinson, Preston
Marfan syndrome

'I drew this Lotus Flower to represent Marfan syndrome because the lotus flower is a deeply meaningful symbol for those navigating chronic illness, like Marfan syndrome, due to its powerful associations with growth, resilience, and transformation. I love doing creative drawings. It helps me express my life with Marfan syndrome.'

More... rewards... than you can imagine

Lorraine Thompson, Huddesfield
DDX3X, X linked neurodevelopmental syndrome

'Leah was diagnosed 2024 with DDX3X syndrome. I Lorraine, now retired, worked as specialist nurse in rare disease field for children and adults for 20 years in Manchester, an approach with two views sometimes. Leah's story is of a beautiful young woman in a life with many health and physical barriers and dedicated parents frustration of having no diagnosis. Leah is dependent on us and has given us more joy than we could have imagined.'

Leah, you're special
In every way
A gift from heaven
We treasure every day
Our family blessed since the day
You were born
Always happy, never forlorn
Brave and resilient
You have proven to be
A source of inspiration
To all the family
Your smiling face
Never blue
A dry sense of humour
Could that be you?
With each new adventure
You continue to astound
So many achievements
What next will be found?
First school, then college
Brownies and Youth Club too
Teachers and helpers
Amazed by what you can do
Always willing to help others
And ready to learn something new
You are a joy to be with
A sentiment so true
The world's greatest shopper
Now that's definitely you!
The latest hi-tech gadgets
That's you too!
You're patient and tolerant
We can all learn from you
'My Life' your latest venture
Social activities galore
Family holidays in abundance
Who could ask for more
So, on your 30th birthday, Leah
Just wanted to say
You're wished every happiness

Enjoy your special day.

The shared experience of rare conditions

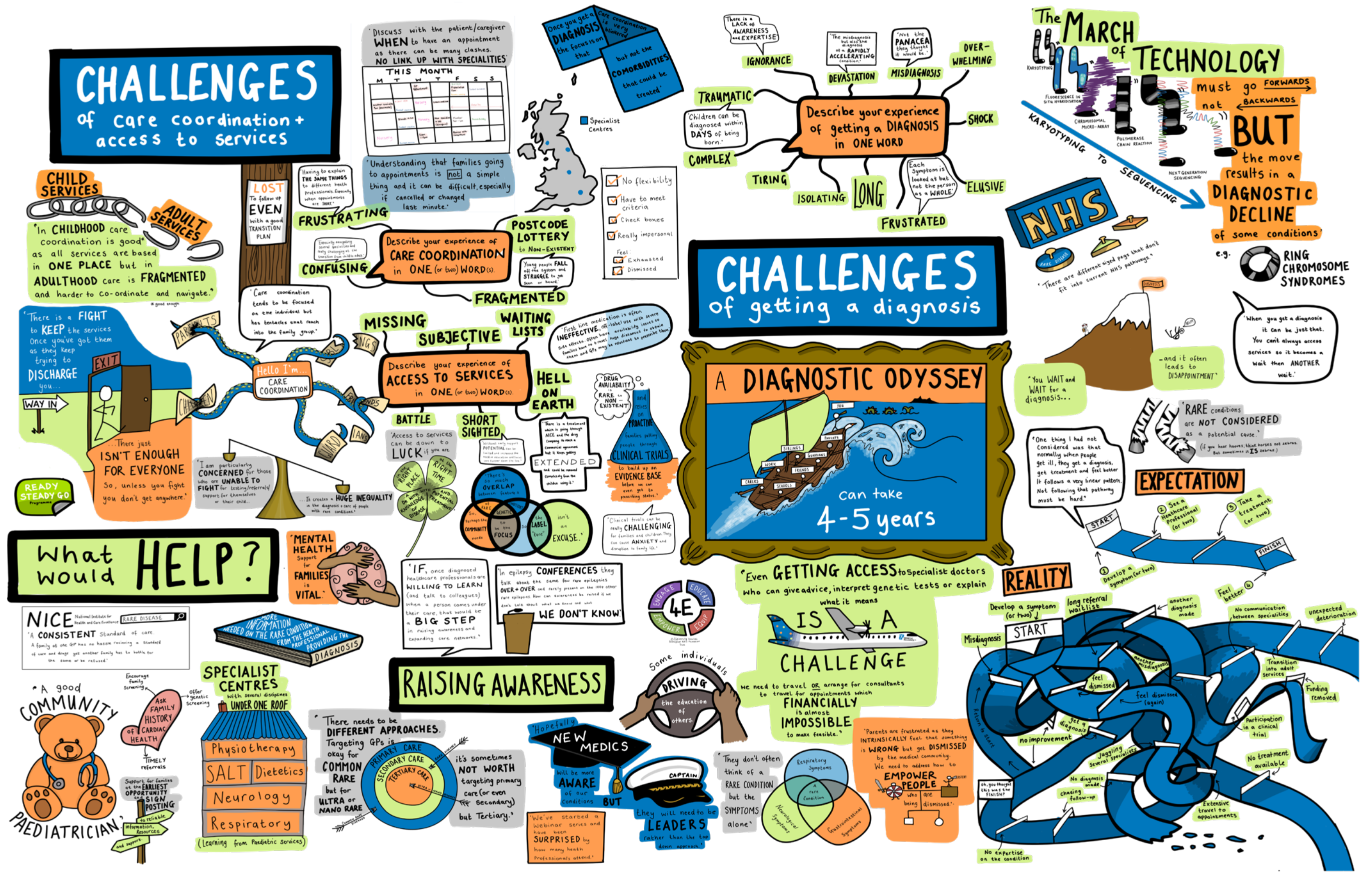
Rose Matheson

Individually, rare conditions are rare but together, there are around 3.5 million people living with a rare condition in the UK.

'More than you can imagine: an anthology of rare experiences' is a collection of creativity showcasing the impact rare conditions can have on a person's sense of self, relationships and wellbeing.

While the anthology shares individual experiences of rare conditions, it is impossible to ignore the shared challenges that underpin each story. Delays in getting an accurate diagnosis, a lack of understanding amongst healthcare professionals, poorly coordinated care and barriers to accessing services and treatment are the common challenges that shape a person's rare experience.

This shared experience has been captured by Rose Matheson (illustrator and paramedic, Scotland). Rose attended Genetic Alliance UK's monthly Member Meeting in January 2025 where members of the rare community shared the most common challenges of living with a rare condition and discussed what would help improve their experience of care. This illustration reflects the key points of that discussion and provides a visual representation of the shared experience of rare conditions.



The tiny zebra trots along
Unaware that she doesn't belong
She is surrounded by her kind
Absent monochrome they don't mind

When hungry lions chase for snacks
They only see the black and white backs
Rainbow zebras are so rare
Beautiful beyond compare